

CLAIMS

What is claimed is:

- 5 1. A method of identifying subjects at risk of developing Crohn's disease comprising:
- a) providing
- i) nucleic acid from a subject, wherein said nucleic acid comprises a Nod2 gene; and
- 10 b) detecting the presence or absence of one or more variations in said Nod2 gene.
2. The method of Claim 1, further comprising step c) determining if said subject is at risk of developing Crohn's disease based on the presence or absence of said
- 15 one or more variations.
3. The method of Claim 2, wherein said determining of step c) comprises determining a genotype relative risk for said subject.
- 20 4. The method of Claim 2, wherein said determining of step c) comprises determining a population attributable risk for said subject.
5. The method of claim 1, wherein said variation is a mutation.
- 25 6. The method of Claim 1, wherein said variation is a polymorphism.
7. The method of claim 1, wherein said variation results in increased NF- B activation.
- 30 8. The method of Claim 5, wherein said mutation is a cytosine residue insertion.

9. The method of Claim 5, wherein said mutation causes a deletion of at least one LRR repeat of Nod2.

5 10. ~~The method of Claim 1, wherein said one or more variations are selected from the group consisting of the nucleic acid sequences described by SEQ ID NOs: 33, 54, 56, 58, 60, 62, 64, 66, 68, 84, 86, and 88.~~

11. The method of Claim 1, wherein said detecting in step (b) is accomplished
10 by hybridization analysis.

12. The method of Claim 1, wherein said detecting in step (b) comprises
comparing the sequence of said nucleic acid to the sequence of a wild-type Nod2 nucleic
acid.

15 13. A kit for determining if a subject is at risk of developing Crohn's disease
comprising:

a) a detection assay, wherein said detection assay is capable of specifically
detecting a variant Nod2 allele; and
20 b) instructions for determining if the subject is at increased risk of
developing Crohn's disease.

14. The kit of Claim 13, wherein said detection assay comprises a nucleic acid
probe that hybridizes under stringent conditions to a nucleic acid sequence selected from
25 the group consisting of SEQ ID NOs: 70-83.

15. An isolated nucleic acid comprising a sequence encoding a polypeptide
selected from the group consisting of SEQ ID NOs: 55, 57, 59, 61, 63, 65, 67, 69, 85, 87,
and 89.

30

16. The nucleic acid sequence of Claim 15, wherein said sequence is operably linked to a heterologous promoter.

5 17. The nucleic acid sequence of Claim 16, wherein said sequence is contained within a vector.

18. A host cell comprising the vector of Claim 17.

10 19. The host cell of Claim 18, wherein said host cell is located in an organism selected from the group consisting of a plant and an animal.

20. An isolated nucleic acid sequence selected from the group consisting of SEQ ID NOs: 54, 56, 58, 60, 62, 64, 66, 68, 84, 86, and 88.

15 21. A computer readable medium encoding a representation of the nucleic acid sequence of claim 20.

20 22. An isolated polypeptide selected from the group consisting of SEQ ID NOs: 55, 57, 59, 61, 63, 65, 67, 69, 85, 87, and 89.

23. A computer readable medium encoding a representation of the polypeptide of claim 22.

25 24. A computer implemented method of determining a patient's risk of developing Crohn's disease comprising:

a) providing:

i) nucleic acid from a patient, wherein said nucleic acid comprises a Nod2 gene; and

30 ii) a computer comprising software for the prediction of a patient's risk of developing Crohn's disease; and

b) detecting the presence of one or more variations in said patient's Nod2 gene to generate genetic variation information;

c) entering said genetic variation information into said computer; and

d) calculating said patient's risk with said software.

5

25. The method of claim 24, further comprising step e) displaying said patient's risk.

10 26. The method of Claim 24, wherein said risk comprises a genotype relative risk.

27. The method of Claim 24, wherein said risk comprises a population attributable risk.

15 28. The method of Claim 24, wherein said variation is a polymorphism.

29. The method of Claim 24, wherein said variation is a mutation.

20 30. The method of Claim 29, wherein said mutation is a cytosine residue insertion.

31. The method of Claim 30, wherein said mutation causes a deletion of at least one LRR repeat of Nod2.

25 32. The method of Claim 24, wherein said one or more variations are selected from the group consisting of the nucleic acid sequences described by SEQ ID NOs: 33, 54, 56, 58, 60, 62, 64, 66, 68, 84, 86, and 88.

30 33. The method of Claim 24, wherein said detecting in step (b) comprises comparing the sequence of said nucleic acid to the sequence of a wild-type Nod2 nucleic acid.